



## spondylothoracic dysostosis

Spondylothoracic dysostosis is a condition characterized by malformation of the bones of the spine and ribs. The bones of the spine (vertebrae) do not develop properly, which causes them to be misshapen and abnormally joined together (fused). The ribs are also fused at the part nearest the spine (posteriorly), which gives the rib cage its characteristic fan-like or "crab" appearance in x-rays. Affected individuals have short, rigid necks and short torsos because of the bone malformations. As a result, people with spondylothoracic dysostosis have short bodies but normal-length arms and legs, called short-trunk dwarfism.

The spine and rib abnormalities, which are present from birth, cause other signs and symptoms of spondylothoracic dysostosis. Infants with this condition have small chests that cannot expand adequately, often leading to life-threatening breathing problems. As the lungs expand in the narrow chest, the muscle that separates the abdomen from the chest cavity (the diaphragm) is forced down and the abdomen is pushed out. The increased pressure in the abdomen can cause a soft out-pouching around the lower abdomen (inguinal hernia) or belly-button (umbilical hernia).

Breathing problems can be fatal early in life; however, some affected individuals live into adulthood.

Spondylothoracic dysostosis is sometimes called spondylocostal dysostosis, a similar condition with abnormalities of the spine and ribs. The two conditions have been grouped in the past, and both are sometimes referred to as Jarcho-Levin syndrome; however, they are now considered distinct conditions.

### Frequency

Spondylothoracic dysostosis affects about one in 200,000 people worldwide. However, it is much more common in people of Puerto Rican ancestry, affecting approximately one in 12,000 people.

### Genetic Changes

The *MESP2* gene provides instructions for a protein that plays a critical role in the development of vertebrae. Specifically, it is involved in separating vertebrae and ribs from one another during early development, a process called somite segmentation. Mutations in the *MESP2* gene prevent the production of any protein or lead to the production of an abnormally short, nonfunctional protein. When the MESP2 protein is nonfunctional or absent, somite segmentation does not occur properly, which results in malformation and fusion of the bones of the spine and ribs seen in spondylothoracic dysostosis.

## **Inheritance Pattern**

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

## **Other Names for This Condition**

- Jarcho-Levin syndrome
- STD

## **Diagnosis & Management**

### Genetic Testing

- Genetic Testing Registry: Jarcho-Levin syndrome  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265343/>

### Other Diagnosis and Management Resources

- Cleveland Clinic: Spine X-ray  
<https://my.clevelandclinic.org/health/articles/spine-x-ray>
- GeneReview: Spondylocostal Dysostosis, Autosomal Recessive  
<https://www.ncbi.nlm.nih.gov/books/NBK8828>

### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

## **Additional Information & Resources**

### MedlinePlus

- Health Topic: Dwarfism  
<https://medlineplus.gov/dwarfism.html>
- Health Topic: Spine Injuries and Disorders  
<https://medlineplus.gov/spineinjuriesanddisorders.html>

### Genetic and Rare Diseases Information Center

- Spondylothoracic dysostosis  
<https://rarediseases.info.nih.gov/diseases/6798/spondylothoracic-dysostosis>

### Educational Resources

- Disease InfoSearch: Spondylothoracic dysostosis  
<http://www.diseaseinfosearch.org/Spondylothoracic+dysostosis/6845>
- KidsHealth: Dwarfism  
<http://kidshealth.org/en/parents/dwarfism.html>
- Nemours Children's Health System: Skeletal Dysplasia  
<https://www.nemours.org/service/medical/skeletal-dysplasia.html?tab=about>

### Patient Support and Advocacy Resources

- Human Growth Foundation  
<http://hgfound.org/>
- International Skeletal Dysplasia Registry, UCLA  
<http://ortho.ucla.edu/isdr>
- Little People of America  
[http://www.lpaonline.org/index.php?option=com\\_content&view=article&id=85](http://www.lpaonline.org/index.php?option=com_content&view=article&id=85)
- Resource list from University of Kansas Medical Center: Dwarfism  
<http://www.kumc.edu/gec/support/dwarfism.html>

### GeneReviews

- Spondylocostal Dysostosis, Autosomal Recessive  
<https://www.ncbi.nlm.nih.gov/books/NBK8828>

### ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22spondylothoracic+dysostosis%22+OR+%22Bone+Dysplasia%22>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28spondylothoracic+dysostosis%5BALL%5D%29+OR+%28Jarcho-Levin+Syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

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